be relevant to you as parents, and to other family members. You will be offered a referral to talk to specialists working in Clinical Genetics

Unexpected result

Sometimes a chromosome change is found that is associated with other health problems not related to the test. In other words, the test may find a chromosome change that does not explain the ultrasound findings but may have implications for the future health of your baby, or other family members. This is called an incidental finding.

Uncertain result

The array may detect changes of uncertain significance, called variants of uncertain significance (VUS). This means that currently there is not enough information available to know whether this change is significant or not. The laboratory may not report these changes. This is because a VUS may not provide reliable information about how your baby will develop or how the pregnancy might progress. A VUS may be reported in some circumstances where further investigations for the baby or genetic testing of family members may help clarify the significance.

Close family relationships

The test might also reveal if a person's biological parents are very closely related to one another This will only usually be reported if the test shows the parents might be first degree relatives. For example, the test shows the biological parents are siblings, or a parent and another child in the family. This type of result can occur even if we are only testing the patient (without samples from parents).

When will I get the results?

The chromosome count of 13, 18 and 21 usually takes 1-3 working days. An array usually takes about two weeks. Some results may take longer. A problem with the sample or test can delay the result. In some cases, the test may fail in which case you will need to discuss your options with your obstetric team.

The obstetric department that has offered you this test will contact you with the results. If a change has been found they will ask if you would like to be referred to talk to specialists in Clinical Genetics.

Where can I get more information?

Antenatal Results and Choices (ARC)
ARC offers information and support to parents before, during and after antenatal screening.
t: 0845 077 2290 / 020 7713 7486
w: www.arc-uk.org

UNIQUE

UNIQUE offers support and information for parents of babies diagnosed with rare chromosome disorders.
t: 0188 372 3356
w: www.rarechromo.co.uk

Your Obstetric contact:
Name:
Job title:
Tel:

AWMGS Genetics Team
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AWMGS
All Wales Medical Genomics Service

Fetal Single Nucleotide Polymorphism (SNP) array test

Patient Information Leaflet

Why is this test being offered to me?

The ultrasound scan of your unborn baby showed anomalies that might be due to a genetic condition. An anomaly is a problem with how the structure of a body organ (such as the heart, brain or bones) has developed. This detailed test is being offered to see if there is an identifiable genetic cause for the anomalies seen on your baby's scan.

Understanding your genetic code

Our genetic code is made up of a series of chemical letters; this is our DNA. We inherit half of our DNA from each of our parents. Our DNA forms thousands of genes which instruct our body on how to grow, function and develop. These genes are packaged into structures called chromosomes.

One way to think about this, is to think of our genetic code as being a library of instructions for making us. In this library, our chromosomes are like bookshelves holding thousands of instruction manuals (our genes).

Usually we have 46 chromosomes in 23 pairs. We inherit one copy of each pair from each parent. The chromosome pairs are numbered from 1 to 22 and the 23rd pair is the sex chromosomes. Girls usually have two X chromosomes (XX) and boys usually have an X and a Y chromosome (XY).

What is a SNP?

We all have variation in our DNA. Most of this does not contribute in any meaningful way to our health or learning abilities. This can sometimes be called natural genetic variation.

Some of this variation is very small and involves only one of the letters in our DNA. These are called SNPs. Different people will have a unique mixture of SNPs.

What is a SNP array test?

This variation allows us to detect thousands of SNPs at various locations along each of the chromosomes. A SNP array shows if there is any chromosome material missing (deleted) or if there is any extra chromosome material present (duplicated). Having too much or too little chromosome material can cause problems with growth, development or cause health issues. It is a very broad genetic test, but will not look for very small genetic changes which might affect how genes work.

How is the test carried out?

A sample of baby's cells is taken by one of two ways;

- A sample is taken from the placenta. This test is called chorionic villous sampling (CVS). This test usually takes place at around 11 to 14 weeks of pregnancy.

- A sample of amniotic fluid (the fluid around the baby) is taken. This is called an amniocentesis. This test usually takes place from 15 weeks of pregnancy.

In both cases, the sample of baby's cells is taken with a fine needle that goes through the mother's abdomen (tummy). This is done very carefully using an ultrasound scan to guide the needle. Both amniocentesis and CVS are associated with a small risk of miscarriage. Your Obstetric doctor can explain these procedures to you in more detail.

A blood sample is also taken from both parents and sent with baby's sample for testing.

What are the advantages of this test?

- This is a detailed test that looks at your baby's chromosomes.
- It might be possible to give you more information about the cause of your baby's anomalies, and what this might mean for their future health and development. This may help guide the management of the pregnancy or delivery and management of the baby.
- It might give you information about the chances of having a baby with similar anomalies in the future.

What are the possible test results?

The initial test will count chromosome pairs 13, 18 and 21. If this test shows a change in the number of any of these chromosomes and it is an explanation for the scan findings, you will be informed of this result and an array will **not** be carried out. If this result is normal then an array test will be initiated. From this you will receive one of four results:

No clinically significant change is found

This means the test did not find a chromosome change that explains the ultrasound scan findings. This result does not exclude a genetic condition in your baby as an array does not detect all genetic changes. Your obstetrician will discuss your future care with you.

A clinically significant change is found

This means that the array finding is likely to have a clinical effect on your baby and may